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DATE: Wednesday, November 19, 2003

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____ 1. Document ID: US 20010039335 A1

L2: Entry 1 of 4

File: PGPB

Nov 8, 2001

Apr 15, 2003

PGPUB-DOCUMENT-NUMBER: 20010039335

PGPUB-FILING-TYPE: new

DOCUMENT-IDENTIFIER: US 20010039335 A1

TITLE: Secreted proteins and polynucleotides encoding them

PUBLICATION-DATE: November 8, 2001

INVENTOR-INFORMATION:

NAME	CITY	STATE	COUNTRY	RULE-47
Jacobs, Kenneth	Newton	MA	US	
McCoy, John M.	Reading	MA	US	
LaVallie, Edward R.	Harvard	MA	US	
Collins-Racie, Lisa A.	Acton	MA	US	
Evans, Cheryl	Germantown	MD	US	
Merberg, David	Acton	MA	US	
Treacy, Maurice	Co. Dublin	MA	IE	
Agostino, Michael J.	Andover	MA	US	
Steininger, Robert J. II	Cambridge	MA	US	
Spaulding, Vikki	Lowell	MA	US	
Wong, Gordon G.	Brookline	CA	US	
Clark, Hilary	So. San Francisco	MA	US	
Fechtel, Kim	Arlington		US	

US-CL-CURRENT: 536/23.5; 435/325, 435/69.5, 530/351

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File: USPT

US-PAT-NO: 6548272

L2: Entry 2 of 4

DOCUMENT-IDENTIFIER: US 6548272 B1

TITLE: Gene encoding for a transmembrane protein

DATE-ISSUED: April 15, 2003

INVENTOR-INFORMATION:

NAME CITY STATE ZIP CODE COUNTRY Shimizu; Nobuyoshi Chiba JP Nagamine; Kentaro Tochigi JP

US-CL-CURRENT: 435/69.1; 435/252.3, 435/254.11, 435/320.1, 435/325, 435/471, 435/71.1, 435/71.2, 530/350

Full Title Unation Front Review (Clarence Units Reference Sequences) effectioned: Clarins Finds Court Ce. (In Spe-

1 3. Document ID: US 6166180 A

L2: Entry 3 of 4

File: USPT

Dec 26, 2000

US-PAT-NO: 6166180

DOCUMENT-IDENTIFIER: US 6166180 A

TITLE: Chromosome 21 gene marker, compositions and methods using same

DATE-ISSUED: December 26, 2000

INVENTOR-INFORMATION:

NAME

CITY

STATE ZIP CODE

COUNTRY

Korenberg; Julie R.

Los Angeles

CA

Yamakawa; Kazuhiro

Los Angeles

CA

US-CL-CURRENT: 530/350

Full Title Litation Front Review Classification trate Reference Sequences Attachments

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L2: Entry 4 of 4

File: USPT

Jun 30, 1998

US-PAT-NO: 5773268

DOCUMENT-IDENTIFIER: US 5773268 A

TITLE: Chromosome 21 gene marker, compositions and methods using same

DATE-ISSUED: June 30, 1998

INVENTOR-INFORMATION:

NAME

CITY

STATE ZIP CODE

COUNTRY

Korenberg; Julie R.

Los Angeles

CA

Yamakawa; Kazuhiro

Los Angeles

CA

US-CL-CURRENT: 435/6; 435/252.3, 435/252.33, 435/320.1, 435/325, 435/348, 435/349, 435/350, 435/352, 435/357, 435/366, 435/372.3, 514/44, 536/23.1, 536/23.5

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1. Document ID: US 6166180 A

L1: Entry 1 of 2

File: USPT

Dec 26, 2000

US-PAT-NO: 6166180

DOCUMENT-IDENTIFIER: US 6166180 A

TITLE: Chromosome 21 gene marker, compositions and methods using same

DATE-ISSUED: December 26, 2000

INVENTOR-INFORMATION:

NAME

CITY

STATE

ZIP CODE COUNTRY

Los Angeles

Korenberg; Julie R. Yamakawa; Kazuhiro

Los Angeles

CA CA

US-CL-CURRENT: 530/350

Full Title Citation Front Review Classification bate Reference Sequences Attachments Larmy 1980 Draw Describings

L1: Entry 2 of 2

File: USPT

Jun 30, 1998

US-PAT-NO: 5773268

DOCUMENT-IDENTIFIER: US 5773268 A

TITLE: Chromosome 21 gene marker, compositions and methods using same

DATE-ISSUED: June 30, 1998

INVENTOR-INFORMATION:

NAME

CITY

STATE

ZIP CODE

COUNTRY

Korenberg; Julie R.

Los Angeles

CA

Yamakawa; Kazuhiro

Los Angeles

CA

US-CL-CURRENT: $\frac{435}{6}$; $\frac{435}{252.3}$, $\frac{435}{252.33}$, $\frac{435}{320.1}$, $\frac{435}{325}$, $\frac{435}{325}$, $\frac{435}{348}$, $\frac{435}{349}$, $\frac{435}{350}$, $\frac{435}{352}$, $\frac{435}{357}$, $\frac{435}{366}$, $\frac{435}{372.3}$, $\frac{514}{44}$, $\frac{536}{23.1}$, $\frac{536}{23.5}$

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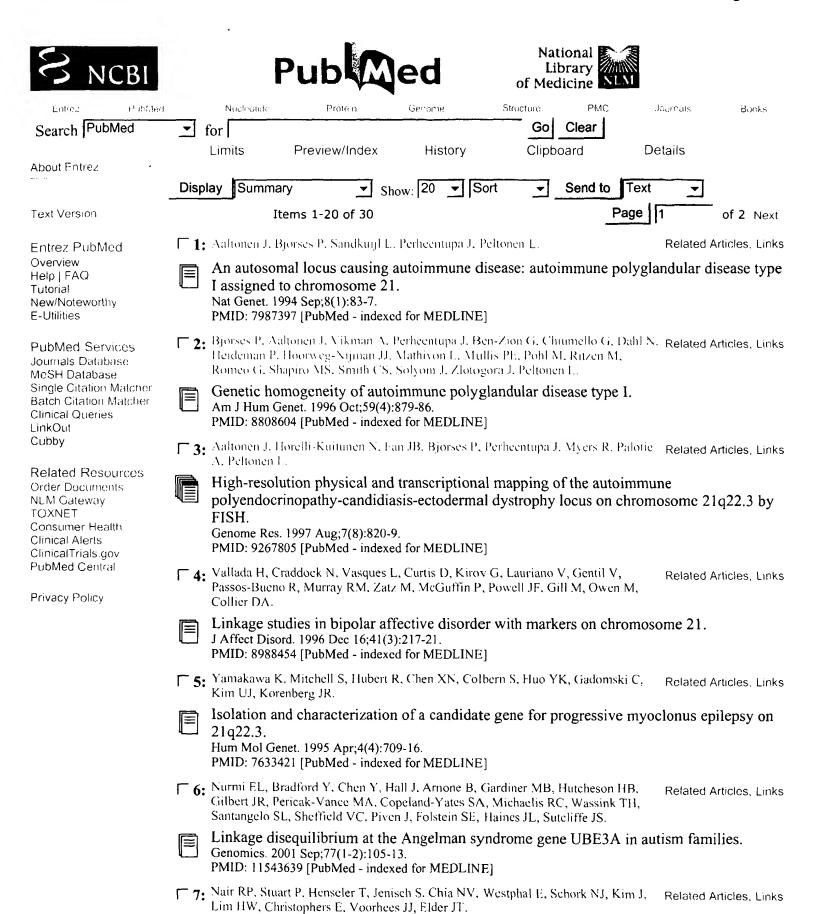
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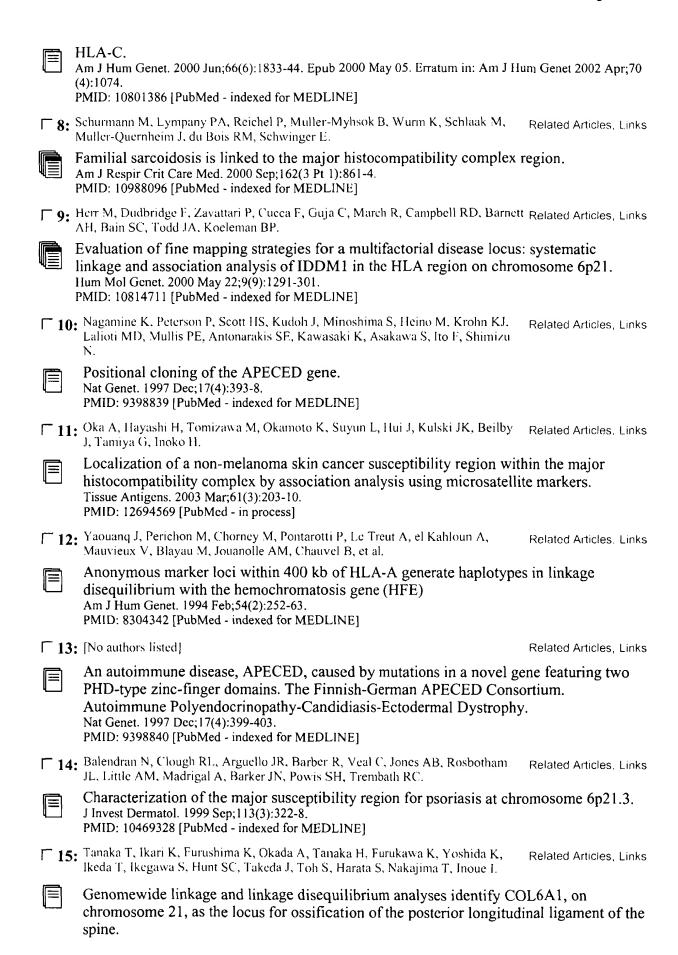
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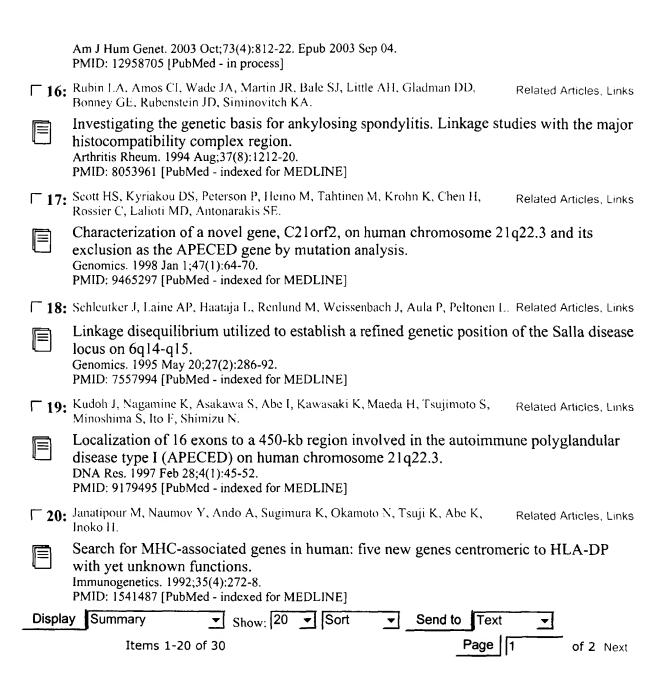
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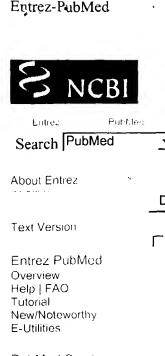
Localization of psoriasis-susceptibility locus PSORS1 to a 60-kb interval telomeric to





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An autosomal locus causing autoimmune disease: autoimmune polyglandular disease type I assigned to chromosome 21.

Aaltonen J, Bjorses P, Sandkuijl L, Perheentupa J, Peltonen L.

Department of Human Molecular Genetics, National Public Health Institute, Helsinki, Finland.

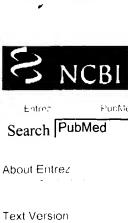
Autoimmune polyglandular disease type I (APECED) is an autosomal recessive autoimmune disease characterized by a variable combination of the failure of the endocrine glands. The pathogenesis of this unique autoimmune disease is unknown; unlike many other autoimmune diseases, APECED does not show association to specific HLA haplotypes. Unravelling the APECED locus will identify a novel gene outside the HLA loci influencing the outcome of autoimmune diseases. We have assigned the disease locus to chromosome 21q22.3 by linkage analyses in 14 Finnish families. Linkage disequilibrium studies have significantly increased the informativeness of the analyses and helped to locate the critical DNA region for the APECED locus to just 500 kilobases, a much more precise definition than linkage analyses alone could achieve.

PMID: 7987397 [PubMed - indexed for MEDLINE]

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	☐ 6: Christin-Maitr	e S, Vasseur C, Portr	noi MF, Bouchard I	Ρ.	Related A	articles, Links				
	Mol Cell Endo	oremature ovarian ocrinol. 1998 Oct 25; 02 [PubMed - indexed	145(1-2):75-80. Re	eview.						
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1. The DNA sequence of human chromosome 21

M. Hattori, A. Fujiyama, T. D. Taylor, H. Watanabe, T. Yada, H.-S. Park, A. Toyoda, K. Ishii, Y. Totoki, D.-K. Choi, E. Soeda, M. Ohki, T. Takagi, Y. Sakaki, S. Taudien, K. Blechschmidt, A. Polley, U. Menzel, J. Delabar, K. Kumpf, R. Lehmann, D. Patterson, K. Reichwald, A. Rump, M. Schillhabel, A. Schudy, W. Zimmermann, A. Rosenthal, J. Kudoh, K. Shibuya, K. Kawasaki, S. Asakawa, A. Shintani, T. Sasaki, K. Nagamine, S. Mitsuyama, S. E. Antonarakis, S. Minoshima, N. Shimizu, G. Nordsiek, K. Hornischer, P. Brandt, M. Scharfe, O. Schön, A. Desario, J. Reichelt, G. Kauer, H. Blöcker, J. Ramser, A. Beck, S. Klages, S. Hennig, L. Riesselmann, E. Dagand, T. Haaf, S. Wehrmeyer, K. Borzym, K. Gardiner, D. Nizetic, F. Francis, H. Lehrach, R. Reinhardt, M.-L. Yaspo

SUMMARY: Chromosome 21 is the smallest human autosome. An extra copy of chromosome 21 causes

Down syndrome, the most frequent genetic cause of significant mental... **CONTEXT: Chromosome 21** represents around 1-1.5% of the human genome. Since the discovery in 1959 that Down syndrome occurs when there are three copies of **chromosome 21** (ref. 1), about twenty disease loci have been mapped to its long arm, and the..... Nature**405**, 311 - 319 (18 May 2000) Article

Abstract | Full Text | PDF

2. Autoimmune regulator: from loss of function to autoimmunity

J Pitkänen P Peterson

SUMMARY: The autoimmune regulator (AIRE) is a gene where mutations cause the recessively inherited disorder called autoimmune polyendocrinopathy–candidiasis–ectodermal dystrophy (APECED) or autoimmune polyendocrinopathy syndrome type...

CONTEXT: ...inherited disease, autoimmune polyendocrinopathy candidiasis ectodermal dystrophy (APECED), also known as autoimmune polyglandular syndrome type 1 (APS-1).1,2 The disease usually Starts with mucocutaneous candidiasis early in...... Genes and Immunity4, 12 - 21 (13 Dec 1901) Reviews Abstract | Full Text | PDF

3. Linkage disequilibrium in isolated populations: Finland and a young sub-population of Kuusamo

Teppo Varilo, Maris Laan, Iiris Hovatta, Victor Wiebe, Joseph D Terwilliger, Leena Peltonen SUMMARY: Linkage disequilibrium (LD), non-random association of alleles at closely linked chromosomal loci, has been used as a tool in the identification of disease alleles,...

CONTEXT: markers in potential study populations can give an idea of the allelic diversity as well as of the background LD, and so offer some indication of the power of linkage analysis, not only for LD analysis, but also for mapping the loci......

European Journal of Human Genetics8, 604 - 612 (01 Aug 2000) Article

Abstract I PDF

4. CTLA-4 in autoimmune diseases – a general susceptibility gene to autoimmunity?

O P Kristiansen, Z M Larsen, F Pociot

SUMMARY: For most autoimmune disorders, the pattern of inheritance is very complex. The major histocompatibility complex (MHC) gene complex has been implicated as the major...

CONTEXT: Genes and Immunity (2000) 1, 170-184 2000 Macmillan Publishers Ltd All rights reserved 1466-4879/00 \$15.00 www.nature.com/gene REVIEW CTLA-4 in autoimmune diseases - a general susceptibility gene to autoimmunity? OP Kristiansen, ZM..... Genes and Immunity1, 170 - 184 (01 Feb 2000) Review

Abstract | PDF

5. 77 C/G mutation in the tyrosine phosphatase CD45 gene and autoimmune hepatitis; evidence for a genetic link

A Vogel, C P Strassburg, M P Manns

SUMMARY: Autoimmune hepatitis is a chronic immune-mediated disease characterized by a loss of tolerance against liver resident antigens. The genetic background of autoimmune hepatitis is. CONTEXT: ...have been studied during the last years, including genes of human leukocyte antigens,2 cytotoxic T-lymphocyte antigen3 and the vitamin D receptor.4,5 In this study, we investigated an association of mutations of the CD45 molecule. Genes and Immunity4, 79 - 81 (13 Dec 1901) Communications Abstract | Full Text | PDF

6. Familial Robertsonian translocation 15;21 and rare paracentric inv(21): unexpected re-inversion in a child with translocation trisomy 21

Ulrike A Mau, Uwe R Petruch, Peter Kaiser, Thomas Eggermann SUMMARY: We present a family with a Robertsonian translocation (RT) 15;21 and an inv(21)(q21.1q22.1) which was ascertained after the birth of a child with Down...

CONTEXT: ...Robertsonian translocation 15;21 and rare paracentric inv(21): unexpected re-inversion in a child with translocation trisomy 21 Ulrike A Mau1, Uwe R Petruch2, Peter Kaiser1 and Thomas Eggermann3 P53 and Ras p21 proteins. In the Æve.. European Journal of Human Genetics8, 815 - 819 (07 Nov 2000) Article

Abstract | PDF

7. A genome-wide scan shows significant linkage between bipolar disorder and chromosome 12q24.3 and suggestive linkage to chromosomes 1p22–21, 4p16, 6q14–22, 10q26 and 16p13.3

H Ewald, T Flint, T A Kruse, O Mors

SUMMARY: The present study reports a genomewide scan using linkage analysis for risk genes involved in bipolar disorder with 613 microsatellite markers including additional testing.

CONTEXT: ...disorder is a severe and common psychlatric disorder with a lifetime risk around 0,3 1.5%. The disease is characterized by affective episodes with manic, depressive and other psychiatric symptoms. Mania and melancholia have been.

Molecular Psychiatry7, 734 - 744 (15 Aug 2002) Original Research Article Abstract | Full Text | PDF

8. Examination of trisomy 13, 18 and 21 foetal tissues at different gestational ages using FISH

G E Moore, P Ruangvutilert, K Chatzimeletiou, G Bell, C-K Chen, P Johnson, J C Harper SUMMARY: In man high levels of aneuploidy are seen in spontaneous abortions. Very few autosomal trisomies survive to birth, the three most common being those...

CONTEXT: ...of trisomy 13, 18 and 21 foetal tissues at different gestational ages using FISH GE Moore1, P Ruangvutilert2, K Chatzimeletiou1,3, G Bell1, C-K Chen2,4, P Johnson1 and JC Harper2 1Department of Maternal **and** Fetal Medicine, Division...... European Journal of Human Genetics**8**, 223 - 228 (01 Mar 2000) Article

Abstract | PDF

9. Paternal meiotic origin of der(21;21)(q10;q10) mosaicism [46,XX/46,XX,der(21;21)(q10;q10),+21] in a girl with mild Down syndrome

Dieter Kotzot, Albert Schinzel

SUMMARY: Mosaicism for a derivative 21, der(21;21)(q10;q10), is a rare chromosomal abnormality. Since a normal cell line is present, mitotic origin is considered. Chromosome examination of...

CONTEXT: ARTICLE Paternal meiotic origin of der(21;21)(q10;q10) mosaicism

[46,XX/46,XX,der(21;21)(q10;q10), + 21] in a girl with mild Down syndrome Dieter Kotzot and Albert Schinzel Institut f"ur Medizinische Genetik, Universit" at Z"urich,.....

European Journal of Human Genetics8, 709 - 712 (01 Sep 2000) Article

Abstract | PDF

10. Numerous potentially functional but non-genic conserved sequences on human chromosome 21

Emmanouil T. Dermitzakis, Alexandre Reymond, Robert Lyle, Nathalie Scamuffa, Catherine Ucla, Samuel Deutsch, Brian J. Stevenson, Volker Flegel, Philipp Bucher, C. Victor Jongeneel, Stylianos E. Antonarakis

SUMMARY: The use of comparative genomics to infer genome function relies on the understanding of how different components of the genome change over evolutionary time....

CONTEXT: The sequence of human chromosome 21 (ref. 8) was obtained from the National Center for Biotechnology Information (NCBI) and aligned with PipMaker to the mouse orthologous sequences (both sequences were hard-masked with Repeatmasker).......
Nature420, 578 - 582 (05 Dec 2002) The Mouse Genome

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